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DNA Fingerprinting

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DNA FINGERPRINTING

An Interactive Qualifying Project Report

Submitted to the Faculty of

WORCESTER POLYTECHNIC INSTITUTE

In partial fulfillment of the requirements for the

Degree of Bachelor of Science

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ABSTRACT

Through researching the evolution of DNA fingerprinting technology, it is concluded that its effects on society are of great importance. This IQP provides a basic background of how DNA profiling is done and the correct methods for obtaining, transporting, and storing DNA forensic samples. Several landmark court cases are discussed showing a progression of acceptance of DNA fingerprinting in the legal system. The purpose, ethics, and privacy rights associated with DNA databases are also described, followed by the authors' conclusions about the influential effects this technology has on society.

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PROJECT OBJECTIVES

The objective of this project was to look at the growing technology of DNA fingerprinting, noting its powerful effect on society. Chapter 1 outlines the main analysis techniques and procedures performed on DNA. Chapter 2 follows with explanations and examples of methods used to collect and store DNA samples gathered as evidence. The purpose of chapter 3 demonstrates how several selected landmark court cases have set legal precedence for accepting complex technological evidence in U.S. courts. The final chapter, 4, describes the two main types of DNA databases (forensic and genetic), and discusses their main purposes along with the privacy rights and ethical concerns that follow. Based on their research, conclusions are made at the end by the authors about this influential yet controversial technology.

Chapter-1: DNA Fingerprinting Technology

Alexandra Beando

Introduction

DNA profiling or fingerprinting is the analysis of DNA from samples of body tissues or fluids to identify individuals. Specific patterns of DNA result from the analysis and are different between individuals. DNA fingerprinting techniques are widely used in areas such as, parentage testing, criminal forensics, and molecular archeology. Identifying individuals is crucial at a crime scene, where DNA can be found in hair, seminal fluid, saliva, skin cells, and blood. The DNA can be used to help convict the guilty or to exonerate the innocent. In parentage testing, DNA can be collected from a simple cheek swabbing of a child and compared to a potential parent. About 99.8% of every individual's DNA is the same, which makes us human. However, about 0.2% of our DNA differs, and its analysis is used for identification purposes. With 3 billion base pairs in each person's DNA, no two individuals have the exact same sequence, except for maybe identical twins (Brinton and Lieberman, 1994), allowing DNA identifications to be obtained if the analysis is properly performed. The purpose of this chapter is to introduce the reader to DNA, its structure and sequences, the repeating domains used for identification, the two main types of DNA analysis, and the main applications for this technology.

Background on DNA

Deoxyribonucleic acid (DNA) is the basis for identification analysis (Collins, 2002). DNA is composed of *nucleotides*, which are composed of a deoxyribose sugar, a phosphate group, and one of four nitrogen bases, adenine, cytosine, guanine, thymine (Brinton and Lieberman, 1994; DNA From the Beginning, 2012). DNA's four types of nucleotides are

assembled into a long bio-polymer (Figure-1) whose structure was originally deduced in 1952 (Watson and Crick, 1953). The DNA molecule is made up of two anti-parallel strands in the form of a double helix, which resembles a twisted ladder, where each parallel strand of DNA is composed of a linear array of nucleotides. The strands are bonded together such that the bases extend toward the central axis of the molecule, and the two backbones (shown as blue in the diagram) are composed of alternating sugar and phosphate subunits. The bases of two strands are weakly bonded to each other using hydrogen bonds in a complementary fashion in which, adenine pairs with thymine, and cytosine pairs with guanine (Use of DNA....2009).

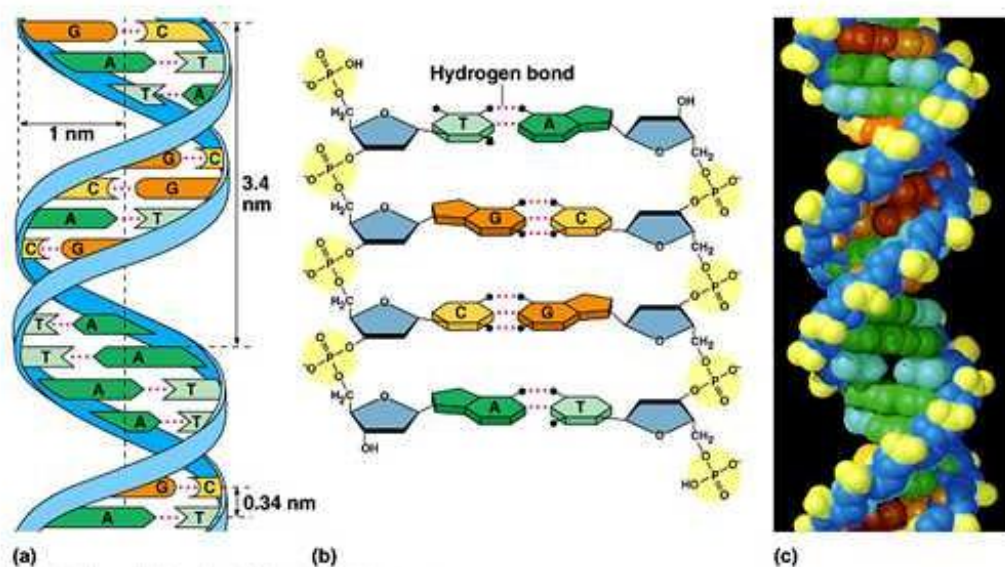


Figure-1: Diagram of the Structure of DNA. Shown in panels A and C are the main double helical structure of DNA. Panel B shows the specific base-pairing between the bases of opposite strands. (Cuny.edu, 2009)

There are two main cellular locations for DNA, and both can be analyzed for profiling purposes. Nuclear DNA resides within the nucleus, while mitochondrial DNA resides within mitochondria. Nuclear DNA is used the most often for forensic analysis (Collins, 2002). Human nuclear DNA exists complexed with proteins in structured called *chromosomes*. Individuals

have forty-six chromosomes in each nucleated cell, with two extra sex chromosomes: XX for female and XY for male (Collins, 2002; DNA From the Beginning, 2012). Each chromosome represents a package for one long strand of nuclear DNA (Figure-2), and the strand is wound around histone cores, which are looped and fixed to specific regions of the chromosome (DNA From the Beginning, 2012).

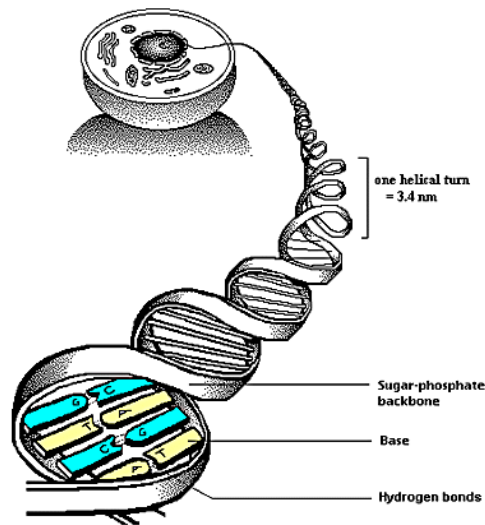


Figure-2: Diagram of a Cell, Nucleus, Chromosomes, and DNA. The diagram shows a long strand of DNA uncoiling from the nucleus of a cell (diagram upper), and the base pairing between the two strands (diagram forefront). (Use of DNA...2009)

When DNA divides (Figure-3), the two parental strands of DNA (left panel) unwind (second panel from left), and enzymes called DNA polymerases use free nucleotides to incorporate onto each parental template (third panel) to make two daughter strands (fourth panel) (Brinton and Lieberman, 1994).

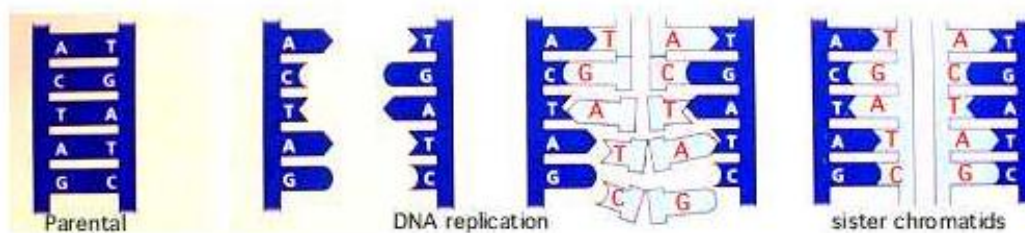


Figure-3: Diagram of DNA Replication. The two parental strands (first panel) separate (second panel) to provide templates for the synthesis of two new daughter strands using free nucleotides (third panel), creating a new daughter strand from each parental template (fourth panel). (Cuny.edu, 2009)

The sequence of nucleotides within DNA acts as genetic information. *Genes* (Figure-4) represent segments of DNA sequences that encode proteins. The sequence of nucleotides dictates the sequence of amino acids in the protein. (Collins, Richard 2002). Genes act as the functional subunits of hereditary information. The collection of genes in an organism determines its characteristics and makes the organism unique (Use of DNA....2009).

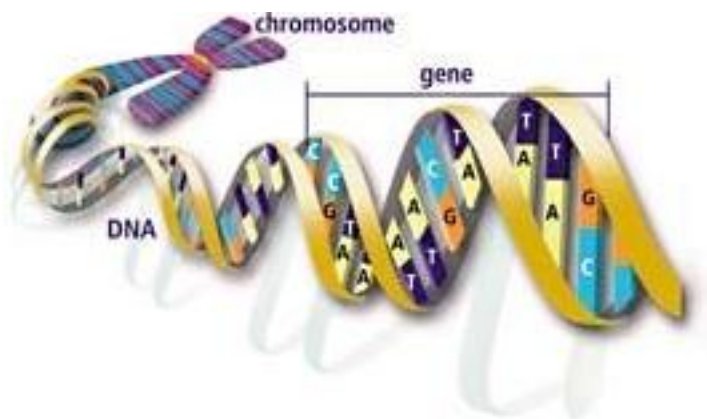


Figure-4: Diagram of Genes on DNA. Genes represent segments of DNA sequences that encode proteins, and are the functional units of hereditary information. (Cuny.edu, 2009)

DNA Loci and Repeating DNA Elements

During DNA fingerprinting, scientists do not sequence an individual's entire DNA sequence, but instead analyze carefully chosen locations on the DNA molecule shown by biologists to differ between individuals. As mentioned previously, most of our DNA has a

conserved sequence, which makes us all human, and this cannot vary much or it becomes non-functional. But 0.2% of our DNA appears to have no functional constraints, and can vary widely between individuals (Brinton and Lieberman, 1994). This DNA often varies by the number of repeating DNA sequences.

There are two main types of repeating DNAs: VNTRs and STRs. Variable number of tandem repeats (VNTRs) (also known as mini-satellites) (Figure-5) are composed of core elements from 9-80 base-pairs long that repeat from 1-30 times at a location (Huskey, 1999; Butler and Reeder, 2004). These repeating elements are relatively long, and are difficult to amplify by PCR (discussed below), so are usually analyzed by RFLP analysis. In the diagram of example VNTRs below, note that an individual can have two different numbers of repeats at one location, each inherited from one parent.

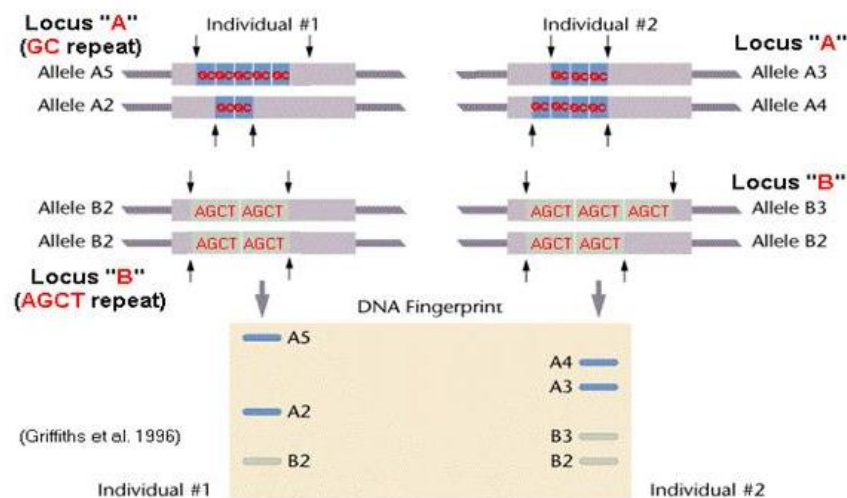


Figure-5: Diagram of Variable Number of Tandem Repeats (VNTRs). Upper panel shows two different individuals (left and right) with a various repeating elements of the dinucleotide GC at locus A, and different AGCT elements at locus-B. For example, individual-1 shows 2 and 5 GC repeats at locus-A, while individual-2 shows 3 and 4 GC repeats at the same locus. At locus B, individual-1 shows 2 repeats of AGCT on both alleles, while individual-2 has 2 and 3 AGCT repeats at the same locus. The lower panel denotes the results of the fingerprint analysis of these VNTRs, two patterns are visible. (Griffiths, 1996)

Short tandem repeats (STRs) are repetitive elements that are shorter than VNTRs, and are easy to amplify by PCR (Butler and Reeder, 2004). Because PCR is sensitive and rapid, the PCR analysis of STRs has become the industry standard for DNA fingerprinting. The Combined DNA Index System (CODIS) is the FBI's DNA database, the world's largest. The current standard for data entry into this database is 13 core STR loci (Butler and Reeder, 2004; Harris, 2010). CODIS was launched in 1998 to attempt to link serial crimes and unsolved cases with the DNA profiles of repeat offenders (Meeker-O'Connell 2004). The FBI database links all fifty states and requires greater than four RFLP markers and/or thirteen core STR markers for identification (Butler and Reeder, 2004). In 2004, the FBI had a backlog of over 600,000 samples that are seeking matches in the system (Butler and Reeder, 2004). If all 13 core loci are analyzed, the likelihood that two individuals will have the same profile is one in a billion, making it an effective means for identifying individuals (Harris, 2010).

Types of DNA Fingerprints

As stated before, when analyzing DNA, an individual's entire DNA sequence is not determined. Instead, specific locations are analyzed known to differ from individual to individual by the number of repeating DNA sequences at that location. DNA fingerprinting technology must be able to distinguish the number of repeating elements at a given location. There are two main types of DNA fingerprinting analysis: non-amplifying RFLPs, and amplifying PCRs.

Non-Amplifying DNA Fingerprints

Restriction enzymes cut DNA at specific sequence locations. For example, the enzyme EcoRI cuts DNA at the sequence GAATTC after the first G residue. If the enzyme cuts DNA twice, a DNA fragment forms. Cutting human DNA with EcoRI makes thousands of EcoRI DNA fragments. Some of these fragments will contain STR loci that differ in length between individuals. Restriction fragment length polymorphism (RFLP) refers to this difference in length between restriction fragments that contain a different number of repeating sequences.

RFLP analysis was the first type of DNA fingerprinting analysis used in 1985 when Alec Jeffreys invented DNA fingerprinting (Jeffreys et al., 1985a). RFLP analysis is actually adapted from a much earlier 1975 analysis by Edward Southern that allows scientists to detect specific DNA fragments from among a complex mixture of DNA fragments by hybridizing the DNA to a labeled probe that is complementary to the sequence of interest (Southern, 1975). RFLP analysis requires a fairly large DNA sample, and can take a week to perform (Harris, 2010). DNA is isolated from crime scene evidence or a suspect, and cut with a restriction enzyme to make DNA fragments. The DNA fragments are then separated by size using electrophoresis in which the DNA is loaded onto a gel and electrodes are placed on the gel so that the positive anode is on the far side of the gel away from the DNA, and the negative cathode is placed on the side of the gel near the DNA. DNA migrates through the gel towards the positive anode due to its phosphate residues that give it a negative charge. The smaller DNA fragment migrate fastest and go the farthest. The pattern of DNA fragments is then blotted to a membrane which is then hybridized to a labeled probe complementary to the specific VNTR of interest. The position of the labeled probe on the membrane is visualized by exposing the membrane to x-ray film (Figure-6). In the figure, the markers in lane-1 and -9 represent a set of DNA fragments of

known decreasing length to allow the size of other DNA fragments to be determined. The DNA of the rape victim is shown in lane-2, and is different than the DNA profile of the rape evidence (lane-3 and -4). Evidence #1 is from a semen stain left on her clothing. Evidence #2 is semen removed from the rape victim. Note that the two evidence profiles match each other, pointing to one perpetrator not two. The control DNA in lane-8 is evidence taken from a previously tested person to be sure the labeled probes performed properly. Note that the DNA profile of the crime scene evidence matches that of suspect-1 (lane-6), but not that of suspect-2 (lane-7), so likely suspect-1 is guilty.

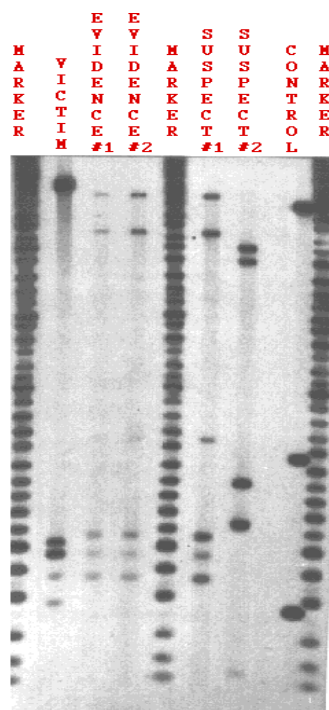


Figure-6: An Example of an RFLP Type Analysis for a Rape Victim. Note how the overall pattern of DNA taken from the rape kit from the victim (lanes 3 and 4) match that of suspect-1 (lane-6) but not suspect-2 (lane-7). (Kimball, 2005)

Amplifying-Type DNA Fingerprints

The second main type of DNA fingerprint is an amplifying type that uses polymerase chain reaction (PCR) to make millions of copies of template DNA, similar to molecular photocopying. It is a fast and inexpensive technique to amplify small segments of DNA, and has become the most popular method of DNA testing when applied to STRs (Rice, 2006). PCR was developed by Kary Mullis in 1986 (Mullis et al., 1986) and earned him the Nobel prize in chemistry in 1993. During PCR, the DNA located between two primers (sense and anti-sense) that flank a region of interest (STR) is amplified, so small amounts of DNA, including from one cell can be analyzed. PCR makes copies of DNA allowing scientists to produce almost any amount to make analyzing the DNA easier. Forensic DNA scientists usually analyze STRs containing tetra-nucleotide repeats, which can easily be amplified using PCRs (Butler and Reeder, 2004). However, throughout the process of PCR the DNA is very sensitive making it prone to possible contamination, any contaminating human DNA is also amplified.

During PCR (Figure-7), the sample of DNA is placed in a test tube containing nucleotides as DNA precursors, sense and anti-sense primers flanking the region of interest, and a special type of DNA polymerase (Taq) that can withstand repeated near boiling temperatures. The tube is placed inside a thermocycler which can be programmed to repeatedly achieve set temperatures. The DNA is initially heated to about 94°C (near boiling) to separate the strands into two pieces of single stranded DNA (step-1 in the diagram). Next, the tube is cooled to about 55°C to allow the primers to hybridize to the DNA (step-2 in the figure). Then the temperature is raised to about 72°C to allow the Taq polymerase to synthesize new DNA using the original strands as templates beginning at the primer sites (step-3 in the figure). This process results in duplication of original target DNA (STR) with each of the new molecules containing one old and

one new strand of DNA (step-4 in the diagram) that can be used to make two more new copies, and so on. The cycle of denaturing, primer annealing, and DNA synthesis is programmed to repeat thirty to forty times, resulting in the production of more than one billion “exact copies” (PCR can make mistakes) of DNA. The entire PCR process can be completed in just a few hours, allowing for scientists to quickly analyze the DNA (Rice, 2006). “PCR has transformed molecular biology through vastly extending the capacity to identify, manipulate, and reproduce DNA. It makes abundant what once was scarce, the genetic material required for experimentations” (Rice, 2006).

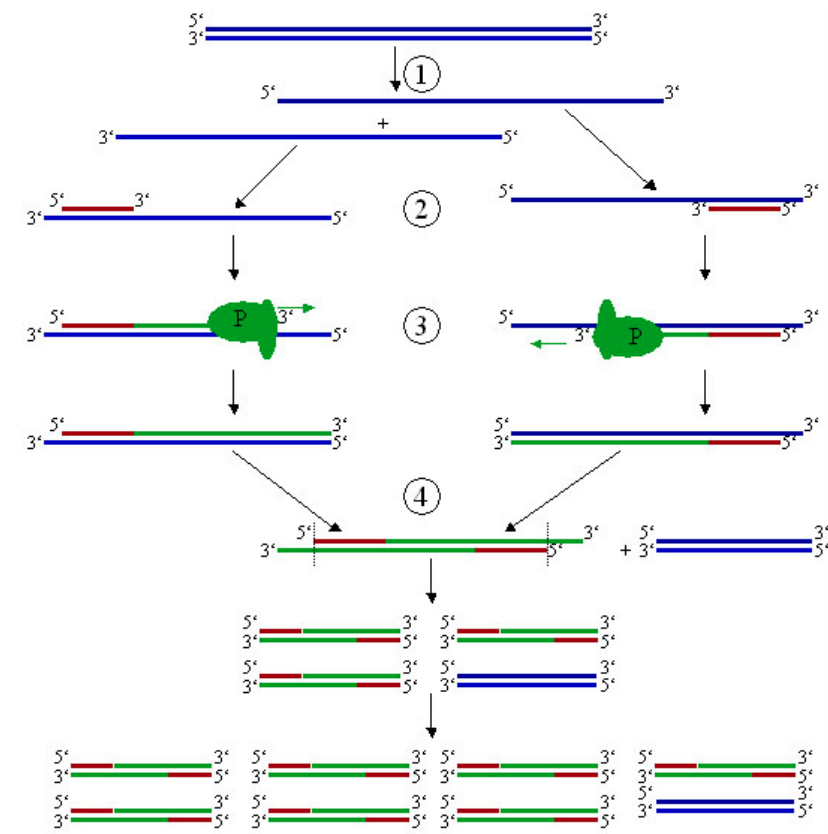


Figure-7: The Main Steps of PCR. Shown in the diagram are the separation of the two template DNA strands (1), the annealing of primers upstream and downstream from the STR locus (2), the synthesis of new DNA catalyzed by Taq polymerase using the primers as start sites (3), and the repeating of the entire process (4). (Rice, 2006)

Once the STR DNA is amplified by PCR, gel electrophoresis or capillary electrophoresis is used to determine the length of the STR band which indicates how many repeat sequences are located at that site. STR analysis examines how many base pairs repeat in specific STR locus on a DNA strand. Analyzing 13 different STR loci allows scientists to accurately identify individuals (Meeker-O'Connell, 2004). STR elements can consist of dinucleotide, trinucleotide, tetranucleotide, or pentanucleotide repeats. Investigators usually analyze tetranucleotide and pentanucleotide repeats to increase sizing accuracy (the longer the repeating element, the greater the length variation which is easier to view) (Harris, 2010).

STR analysis is replacing the traditional RFLP analysis because it requires a smaller sample of DNA (Harris, 2010). PCR-based STR analysis has several advantages over conventional blotting techniques for VNTRs, making STR analysis a more effective means of analysis. Their smaller size is strongly amplified by PCR (so the quantity and integrity of the DNA sample is less of an issue), STRs are highly heterozygous (showing a different number of repeats on the version of the STR inherited from the mother and father, which increases their identification usefulness), and the repeat length is regular (once the repeat length has been established at fertilization, it does not vary in that individual) (Butler and Reeder, 2004).

DNA Fingerprinting Applications

Paternity Testing

Paternity testing was the very first use of DNA fingerprint technology (Jeffreys et al., 1985b). DNA testing has made the process of parentage testing convenient and accurate making it the main application for this technology. Around 220,000 paternity tests are done annually, usually for financial reasons when child payments are required (Health and DNA, 2006). The

applications of parentage testing include the right to abode applications, immigration problems, child support claims, and establishing rights to assets (Collins, 2002). Courts allow the technology when performed properly to establish legal responsibility and child support to parents, while fixing emotional and social issues resolved by knowing the father which in turn increases bonding and active partaking in each other's lives (Health and DNA, 2006). Parentage testing can be completed with the DNA from a cheek swab (Collins, 2002).

The test results in parentage testing are usually more straightforward than criminal forensics, because in parentage testing usually all three parties are known (for example known mother, potential father, and offspring) (Health and DNA 2006). The results of paternity testing can be classified as exclusion or inclusion (Figure-8). In a paternity exclusion, shown in the left panel of the figure, the alleged father's DNA (lane-4) does not match the child, eliminating him from being the father. In a paternity inclusion (right panel), the alleged father (lane-4) shows one STR band (his lower band) whose length matches an STR in the child, so he is a possible parent. By analyzing more STR loci, a more definitive conclusion can be reached about whether more STR loci in the father also match the child.

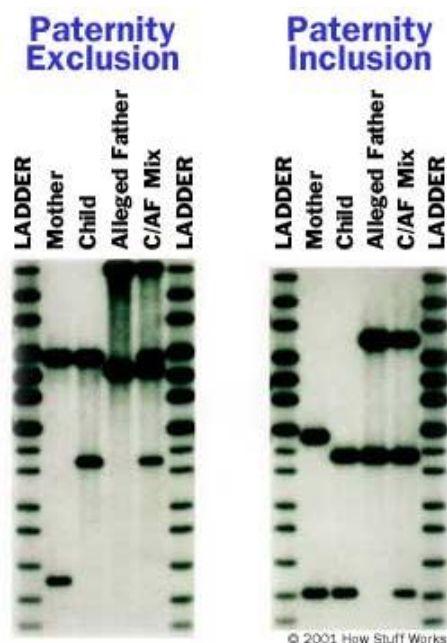


Figure-8: Photograph of a DNA Paternity Analysis. The figure shows two types of paternity analyses, an exclusion (left panel) and an inclusion (right panel). In the exclusion, the alleged father (lane-4) has a DNA profile different than the child. In the inclusion analysis, the alleged father shows a band in common with the child, so might be a parent. (Harris, 2010)

DNA Forensics

Perhaps the best known use of DNA testing is to help solve crimes. Processing a crime scene is a long and tedious process (discussed in Chapter-2) that requires purposeful documentation of the DNA evidence, and the use of methods to avoid its degradation or contamination. All individuals on the scene should be properly trained to preserve the evidence and scene in its original form for subsequent analysis. The DNA profiles from crime scene evidence are compared to entries in a DNA database, such as the previously mentioned FBI's CODIS, which contains profiles from previous offenders and from other crime scenes. Matching profiles between crime scenes has helped identify serial killers or rapists for example. Matching the profile from a current crime to a database of previous offenders helps identify the individual (Layton, 2004). Each state has logged thousands of entries into the CODIS database, helping to link criminals to multiple crimes (Harris, 2010).

Molecular Archeology

Molecular archeology is a field of science that studies the evolution, migrations, and histories of human populations. For example, scientists can use DNA samples extracted from ancient skeletons and compare them to present day people to show how early human populations may have migrated across the globe and to show where they likely came from (Meeker-O'Connell, 2004). As another example, DNA analysis on the mitochondrial DNA of the 5,000 year old mummy Otzi the iceman, showed that he likely originated from a small village in the Italian alps (Ermini et al., 2008).

Other uses for DNA fingerprinting include identifying the remains of unknown soldiers. Individuals serving in the military are required to provide a DNA sample whose profile is put

into a system as a backup for dog-tags while fighting in war. Scientists are also using DNA fingerprinting to match family members with bodies taken from the world trade center disaster (World Trade Center....2001).

Chapter-1 Conclusions

Since its origins in 1985, DNA fingerprinting analysis has become the most powerful technology for identification purposes. Of the two main ways to perform DNA fingerprinting, the PCR-STR technique has become the industry standard due to its sensitivity and speed. DNA analysis is most frequently used for paternity testing, but is also used to help solve crimes, map human origins, and to identify human remains.

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CHAPTER-2: DNA FORENSICS

Gabrielle Demac

Since the discovery of DNA fingerprinting in 1985 (Jeffreys et al., 1985a), society has slowly become more involved and accepting towards the use of DNA evidence in the courtroom. The previous chapter discussed the main ways in which DNA fingerprinting analysis is performed. But the best technology is useless in the courtroom if the DNA evidence was not collected, transported, and stored correctly to prevent contamination and degradation. Proper procedures for properly handling DNA evidence have developed over the years in response to court cases in which the DNA evidence was disallowed. The purpose of this chapter is to discuss some of these procedures, with a focus on gaining evidence admission in courts.

Improvements in crime scene control, evidence collection, and prevention of DNA degradation or contamination, have all contributed to this flourishing field. What began as a test for paternity (Jeffreys et al., 1985b), has turned into technology to help solve major crimes, and aid us in learning about where we are headed as a human race. Without proper collection, transport, maintenance of the samples, and chain of custody to prevent tampering, DNA can potentially become altered, resulting in false reports and accusations of the wrong suspect(s).

Securing the Crime Scene

Let's start at the most important place to collect any piece of evidence, the crime scene. Basic steps to control the crime scene are essential in reducing potential contamination, and is the responsibility of the first officer on the scene. Most investigators follow a layered series of zones (Figure-1) where a primary zone is established immediately around the crime, followed by

other secondary zones surrounding it. The three stages for processing a scene in an organized matter are scene recognition, documentation and collection (Byrd, 2000). Scene recognition is the first task; and by examining the scene and creating different levels of “containment,” police and investigators are able to significantly reduce the risk of intruders such as media and civilians, which reduces the risk of loss or contamination of valuable evidence (Dagnan, 2006).

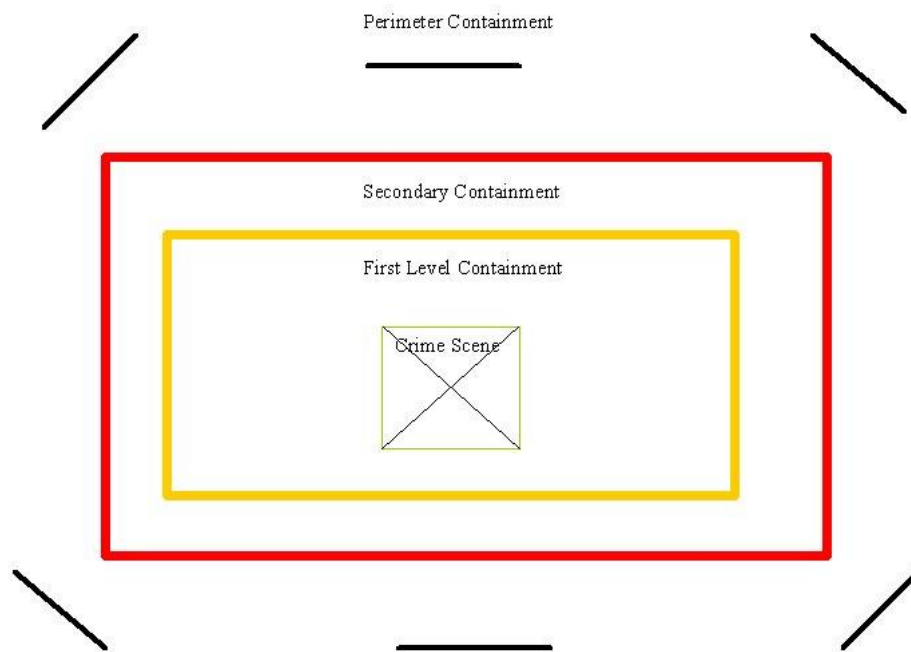


Figure-1: Diagram of “Multi-Level Containment” as an Effective Way of Securing a Crime Scene. (Dagnan, 2006)

Once the scene is secured, with various established perimeters, detailed documentation occurs through sketches, notes, and photography soon followed by the collection of evidence.

Identifying DNA Evidence

Before methods of extracting various types of DNA are discussed, we must first look at where DNA is usually found. DNA is considered to be the “blueprint for life”, and is found in every cell in the body, except red blood cells (DNA...2003). At crime scenes, DNA is most commonly collected from blood, semen, skin cells, tissue, organs, muscle, brain cells, bone, teeth, hair, saliva, mucus, perspiration, fingernails, urine and feces (Evidence Collection...2002). For cold cases, or older crimes that have not been solved yet, DNA can be extracted from bone or teeth, as they do not degrade nearly as quickly as bodily fluids or tissue. Table-1 contains example tissues, their DNA content, and typical PCR success rates. Note that blood (20,000-40,000 ng/ml) and semen (150,000-300,000 ng/ml) are the best sources of DNA. Also note that poor PCR success is obtained from DNA left on handled objects like a door knob, compared to biological fluids, so the latter is preferred if possible.

Table-1: DNA Content and PCR Success Rates of Various Biological Fluids
(Kaye and Sensabaugh, 2000)

Type of Sample	DNA Content	PCR Success Rate
Blood	20,000-40,000 ng/mL	
-stain 1 cm x 1 cm	Ca. 200 ng	>95%
-stain 1mm x 1 mm	Ca. 2 ng	
Semen	150,000-300,000 ng/mL	
-on post coital vaginal swab	0-3000 ng	>95%
Saliva	1000-10,000 ng/mL	
-on a cigarette butt	0-25 ng	50-70%
Hair		
-root end of pulled hair	1-750 ng	>90%
-root end of shed hair	1-12 ng	<20%
-hair shaft	0.001-0.040 ng/cm	
Urine	1-20 ng/mL	
Skin Cells		
-from socks, gloves or clothing repeatedly used		30-60%
-from handled objects (e.g doorknob)		<20%

It is also very helpful for police and investigators to know in advance common items that may potentially contain DNA evidence. Table-11 lists common pieces of potential evidence, and lists where the DNA may be found on that item, and the expected biological source of the cells providing that DNA. Obviously, many other possible locations for DNA can be found, but this list shows the most common an officer is most likely to encounter.

Table-2: Common Pieces of Physical Evidence Containing DNA.
(What Every...1999)

Evidence	Possible Location of DNA on the Evidence	Source of DNA
baseball bat or similar weapon	handle, end	sweat, skin, blood, tissue
hat, bandanna, or mask	inside	sweat, hair, dandruff
eyeglasses	nose or ear pieces, lens	sweat, skin
facial tissue, cotton swab	surface area	mucus, blood, sweat, semen, ear wax
dirty laundry	surface area	blood, sweat, semen
toothpick	tips	saliva
used cigarette	cigarette butt	saliva
stamp or envelope	licked area	saliva
tape or ligature	inside/outside surface	skin, sweat
bottle, can, or glass	sides, mouthpiece	saliva, sweat
used condom	inside/outside surface	semen, vaginal or rectal cells
blanket, pillow, sheet	surface area	sweat, hair, semen, urine, saliva
"through and through" bullet	outside surface	blood, tissue
bite mark	person's skin or clothing	saliva
finger nail, partial finger nail	scrapings	blood, sweat, tissue

Extracting DNA From Evidence

Extracting DNA from different evidence sources (substrates) requires different methods and precautions that must be upheld or the sample becomes contaminated or degraded. Since only small samples of DNA are sometimes found, the risk of contamination during the processes of identifying, collecting, and processing the evidence increases dramatically. Gloves should be worn at all times, and changed frequently when handling anything that could be a potential biohazard or is open to contamination, since they both protect the examiner and decrease the risk of cross contamination. Depending on what type of sample is being collected, proper instruments should be used and disposed of, or cleaned thoroughly both before and after handling each individual sample. Because of technological advances such as PCR that are extremely sensitive, investigators should avoid talking, coughing, sneezing, and touching the area believed to hold the DNA to avoid cross contamination. And with PCR, contaminating DNA is likely to be amplified along with the crime scene DNA, so DNA sources cannot easily be distinguished from one another, and if contaminated, could result in incorrect information used in court. When packaging the samples, they should be air dried and stored into new, un-used paper containers (which vary depending on the size of the sample); evidence samples containing DNA should not be stored in plastic as any evidence that is “wet” or contained in plastic runs a high risk of degrading the DNA, since “moisture allows the growth of microorganisms that can destroy or alter evidence” (Farr, 2008).

Blood evidence is especially useful at a crime scene as it contains large amounts of DNA and it is present at a variety of crimes. Blood evidence plays strong roles in crimes such as homicides, burglaries, assaults, etc. Tests can determine whether it is human or animal, its age,

sex, and blood type (Farr, 2008). Communication between the first responder at the scene of the crime, the case detective, and the forensic scientist is also crucial to effectively process the evidence (Farr, 2008). Blood stain analysis has three open categories: 1) Conventional Serological Analysis, 2) RFLP (restriction fragment length polymorphism) Analysis, and 3) PCR (polymerase chain reaction) Analysis. The first category poses the greatest risk of degradation due to its analysis of the proteins (enzymes and antigens) which degrade quickly at a crime scene. The second and third categories analyze DNA sequences in white blood cells. While PCR is beneficial and only requires a small sample, RFLP is especially “statistically individualizing” and therefore more reliable and valid in court, even though it requires a larger sample size.

When collecting blood evidence, it should generally be contained in clean paper packets, envelopes, or bags. However, the type of blood sample determines specifically how it should be obtained. For example, small dried bloodstains are packaged right away, minimizing contamination, while larger dried bloodstains require a portion of the stained item to be cut out along with a clean control sample piece away from the stain. Dried bloodstains can also be tape lifted (a simple and minimizing process) scraped, or absorbed. The latter two methods are less effective and hold a higher risk of contamination. Wet bloodstains collected are generally favored since more tests can be run, so they are handled with priority and slightly differently (Farr, 2008).

One major precaution that should be noted in any investigation is the use of luminol, a “visualization enhancing chemical” (Farr, 2008). Sometimes at the crime scene, it appears to the naked eye that there is no blood present, but as forensic scientists know, “nothing vanishes without a trace” (Harris, 2005). The purpose of luminol is to reveal small, seemingly invisible traces of blood by creating a chemical reaction with blood to make it visible. A reaction occurs

between added chemicals and the hemoglobin in blood to create a blue-green glow from the blood that was left behind (Figure-2). But while luminol can prove quite useful for detecting invisible blood samples, and help investigators note blood patterns, which aid in determining point of attack and maybe even weapon of choice, luminol also proposes some problems. Not only does luminol react with hemoglobin causing blood to glow, it can also potentially react with common household items such as bleach, which may lead investigators down a false road. And most importantly, luminol's chemical reaction damages DNA through the loss of many genetic markers (Farr, 2008). So, luminol is only used after most other options have been exhausted (Harris, 2005).



Figure-2: The Use of Luminol at a Crime Scene. The figure shows the before (left) and after (right) use of luminol, where before there appears to be no sign of blood, but after the chemical reaction occurs the glow reveals the traces (Harris, 2005)

DNA Transport and Storage

Once evidence samples are collected, the next important step is preserving them during transport, storage, and analysis. After being secured in their proper containers, samples containing DNA should be kept dry and at room temperature during transportation and initial storage. Direct sunlight is potentially damaging to DNA, so prompt transportation of evidence is crucial (What Every...1999). Each package should be properly sealed and labeled with the investigator's initials, date, time and location of collection, and a full description of where the evidence was found (Farr, 2008). This labeling begins a sample's 'chain of custody', which documents every person who has handled that specific piece of evidence. Failure to complete any part of this labeling could result in false accusations due to contamination, tampering, or sample mix-ups. For long term storage, stains and controls should be kept frozen after initial use to prevent damage during repeated thawing. Any liquid evidence such as blood or saliva should be refrigerated, not frozen or stored in any type of plastic container since, again, any type of moisture can cause DNA degradation (Byrd, 2000).

Chapter-2 Conclusion

With all the advances in technology to obtain, process, and utilize DNA, over the years the public has grown more accepting towards the use of DNA in the courtroom, and on many occasions, DNA forensics has proven its ability and strength. When all procedures are carefully followed, and DNA is successfully collected without contamination or degradation, that evidence is considered to be one of the most powerful and reliable sources of evidence in the courtroom. It not only places a specific person or persons at the crime scene, but it also helps to link

weapons and other evidence together, or can even link separate crimes together, making the crime more easily solved.

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Chapter-3: Landmark DNA Court Cases

Alexandra Beando

When applying DNA fingerprinting to society and its court cases, this powerful identification technology has gradually become extremely useful in cases where DNA evidence is a main factor in the outcome of the trial. As DNA testing gradually became accepted in U.S. courts, judges must now hold a pretrial hearing to determine whether the methodology is generally accepted in the scientific community, reliable, done properly with controls, and more probative than prejudicial. The purpose of this chapter is to describe the landmark court cases that now mandate these pre-trial hearings for DNA evidence.

Introduction to Landmark Cases

Any time new scientific technology is introduced in the courtroom as testimony, it must pass certain tests of acceptability in the scientific community, which is discussed prior to trial (Ramsland, 2003). By doing so, the court hopefully is able to avoid evidence that is faulty, contaminated, degraded, or the methodology not performed properly to be reliable. Many things can happen to a DNA sample between its collection, transport, storage, PCR-STR analysis, and final interpretation, making DNA evidence now judged on a case-by-case basis (Ramsland, 2003). U.S. courts now usually accept the newer 13 core loci techniques for accurately analyzing DNA evidence due to their ensured reliability. If a match occurs, a statistical analysis is performed to determine the probability that the match could have occurred randomly. This statistical result is used by juries to help determine whether a suspect is guilty or innocent. No U.S. court has rejected DNA evidence on the grounds that the underlying scientific theory that

every individual's DNAs are different, and that DNA testing can in theory visualize this difference, however, some courts have excluded it as evidence because of problems with the possible contamination of samples, questions surrounding the significance of its statistical probabilities, and laboratory errors. States are free to adopt their own standards for the admission of evidence, and most states have passed laws that recognize DNA evidence as admissible in criminal cases, while others have enacted laws that allow DNA evidence to help resolve civil paternity cases (The Gale Group, 1998). Below, six key landmark cases are described that helped define the current methods for allowing complex technical information into U.S. courts.

Frye v. US, 1923

On November 25, 1920 Dr. Robert W. Brown was shot in his office in Washington DC in the evening, while another physician was in the office and was able to witness the shooting (Fisher, 2008). A young black man, named James Frye, had shot and killed the wealthy physician. As Frye ran out of the office, he fired at the witness who attempted to stop him (Fisher, 2008). Because of this, the witness did not recognize Frye, leaving the police with no suspect for the crime. Seven months later, Frye committed an armed robbery, which led to his arrest on August 21, 1921 (Fisher, 2008). When questioned, Frye confessed to the robbery, and also admitted murdering Dr. Brown (Fisher, 2008). The court appointed Frye with a defense attorney, Richard V. Mattingly, who immediately advised Frye to take back his confession. Frye suddenly came up with an alibi, claiming he had been visiting a woman named Essie Watson (Fisher, 2008). William Marston was called into the case by Mattingly a few weeks before the trial because no witnesses to support Frye's claims would come forward. Marston went to jail to visit Frye on June 10, 1922 and gave him a systolic blood pressure test, which is a precursor to

the modern lie detector test. The test involved Marston monitoring the suspect's blood pressure while asking him a series of questions. The method used a standard blood pressure cuff and a physician's stethoscope. The test indicated that Frye was 'innocent', but the defense was unable to prove the crude test was generally accepted in the scientific community (Fisher, 2008). The trial began on July 17, 1922 in a Washington DC court before Judge William McCoy. The defense was based on the lie detector result and Frye's own alibi. The judge did not allow Marston's to take the stand to explain his lie detector test or to do a demonstration in court, but a debate about the validity of the test was done in front of the jurors (Fisher, 2008). Frye's defense fell apart, making the trial last only four days. In those four days, Frye testified that someone had scared off all of his witnesses (Fisher, 2008). The argument of the admissibility of Marston's blood pressure test was done in front of the jury, therefore the jurors knew that Marston's results supported his innocence, but they agreed with the judge that the test was not proven. The jury deliberated for three hours and came to a guilty verdict (Fisher, 2008). Frye avoided the death penalty by being convicted of second degree murder rather than first, mostly due to the debate about the lie detector test. Instead, he was sentenced to life in prison; the knowledge of the lie detector test had influenced the jury, although it was not part of the case, nor deemed proven. Judge McCoy was trying to avoid this reaction from the jury by not allowing the blood pressure evidence in court if it was unproven technology.

Defense attorney Mattingly appealed Frye's earlier conviction in the District Court on the grounds that Marston's blood pressure test was not allowed in court (Fisher, 2008). In 1923, the Circuit Court of Appeals in the District of Colombia agreed with Judge McCoy's District Court earlier exclusion of the lie detector evidence in court, thereby establishing a new test that became known as the Frye Standard for admitting expert testimony and technology based on *generally*

accepted scientific principles (Fisher, 2008). According to the Appellate Judges, the systolic blood test administered to Frye was not up to the standards of scientific researchers (Fisher, 2008). In a unanimous decision, the three-judge appellate court ruled for the United States in a short opinion that became one of the most famous opinions written by a federal appeals court (*Frye v. United States 2010*).

Senior Appellate Judge Van Orsdel set the general standards for admitting scientific evidence that stood for the next 80 years (Adler, 2008; Fisher, 2008). The polygraph test itself however was not the principle that Judge Van Orsdel was against. This continued to cause issues and controversy with subsequent polygraph tests for over the next sixty years (Fisher, 2008), and lie detector evidence has been largely excluded from criminal trials to the present day, however it is allowed to be used in other settings. The judge stipulated that experts can only testify about scientific matter that has gained general acceptance in the particular field in which it belongs (Adler, 2008).

After serving 18 years in prison, Frye was paroled from the District of Colombia Prison at Lorton, Virginia on June 17, 1939. Judge McCoy's and Van Orsdel's decisions became an important and historical legal precedent for determining admissibility of technical evidence. This Frye case did not involve DNA evidence, however it set a basis for later standards involving DNA evidence in the courtroom.

Sarbah v. Home Office, 1985

Andrew Sarbah was a British citizen who had spent eleven years in Ghana with his father. While trying to get back into England, he was detained by immigration officials at Heathrow due to a suspicion of a forged passport. Sarbah's lawyer was aware of a new DNA

technique pioneered by Alec Jeffreys, and contacted Jeffreys to prove that Sarbah was a British citizen. Jeffreys could not get a DNA sample from Andrew's father or aunts, but he got samples from his mother, brother, and two sisters. The test results performed by Jeffreys confirmed Sarbah was related, and he was allowed to stay in England (Jeffreys et al., 1985). DNA fingerprinting allowed for Sarbah to get back into England by proving his relatives already resided there. This was the first case in the world to use DNA fingerprinting (Jeffreys et al., 1985).

Colin Pitchfork, 1987

In England, two connected murders occurred, one in Narborough, Leicester in November 1983, and the other in Enderby, Leicester in July 1986. On November 21, 1983, 15-year-old Lynda Mann left her house to visit a friend and never returned home. The next morning she was found raped and strangled on a deserted path known as the Black Pad. Investigators used crude enzymatic typing on a semen sample taken from the victim. The crude enzyme profile created in the analysis only matched ten percent of males with blood type A. With no leads or evidence aside from the enzymatic profile, the case was left open (Colin Pitchfork, 2007).

On July 31, 1986 15-year-old Dawn Ashworth took a shortcut instead of taking her normal route home. Two days later her body was found in a wooded area near a footpath called Ten Pound Lane. She was beaten, raped, and strangled to death, just as Lynda Mann had been eight months prior. But this time, a more sophisticated DNA profile was created for the case. Investigators believed the prime suspect was seventeen-year-old Richard Buckland, because he revealed the body of Ashworth to the police. He admitted to the crime through questioning, but denied the first murder of Lynda Mann. As DNA profiling was recently created by Alec Jeffreys and Peter Gill, it could be used to help identify the criminal and solve the murder cases. Gill

commented, “The biggest achievement was developing the preferential extraction method to separate sperm from vaginal cells – without this method it would have been difficult to use DNA in rape cases,” (Colin Pitchfork, 2007). Jeffreys compared semen samples obtained from both murders against a blood sample given by Buckland. This analysis proved that both girls were killed by the same man, however that man was not Buckland. Thus, Buckland became the first person to have his *innocence* proven through DNA fingerprinting. Jeffreys said, “I have no doubt whatsoever that he would have been found guilty had it not been for DNA evidence. That was a remarkable occurrence,” (Colin Pitchfork, 2007). So, Buckley was the first murder exoneration using DNA testing.

With no new leads, the investigation set a precedence as five thousand local men were asked to donate blood and saliva samples for a screen to compare their DNA with the victims, making this case the first to use mass DNA screening. But after six months of analysis, no matches were found and the case seemed as though it may never be solved. Then in a bar, a woman overheard a man named, Ian Kelly, bragging about receiving two hundred Euros for giving his blood for his friend Colin Pitchfork, a local banker (Colin Pitchfork, 2007). Authorities then tested Pitchfork, whose DNA was found to match both crime scenes. He confessed, and was arrested at his home in Haybarn Close. Pitchfork then admitted to flashing females over a thousand times, which led to several sexual assaults. Pitchfork strangled his victims to protect his identity and not get caught. In trial, he pleaded guilty to the rape and murders of Mann and Ashworth, and was sentenced to life in prison on January 22, 1988.

On May 14, 2009 Pitchfork’s legal appeal was heard at the Royal Courts of Justice in London. He won a two year reduction in his original sentence of thirty years minimum, making him eligible for release in 2016 (Colin Pitchfork, 2007). The Lord Chief Justice stated that

Pitchfork “cannot be released unless the safety of the public is assured.” While in prison Pitchfork is attempting to improve his character by converting sheet music to brail, and hopes to work with and help the blind when released (Colin Pitchfork, 2007). Pitchfork was the first person to be *convicted* of murder based on DNA fingerprinting evidence, and the first to be caught as a result of mass DNA screening.

People v. Castro, 1989

On February 5, 1987, Vilma Ponce and her two year old daughter were stabbed to death in the Bronx, New York. Police questioned Jose Castro a handyman in the neighborhood and a bloodstain was noticed on his watch during questioning. The DNA from the victims and Castro were analyzed at Lifecodes in Valhalla, New York and the results confirmed a match between the samples implicating Castro. To determine whether the DNA evidence would be allowed at trial, a Frye hearing was held before Judge Scheindlin of the Superior Court of Bronx County, New York, where the prosecution anticipated using the DNA results in court as evidence against Castro (Patton, 1990). But during the hearing the defense attorney questioned whether the Frye standard was through enough for assessing DNA evidence, and in particular did not address whether the samples were analyzed properly with controls.

The judge agreed, and DNA testing science underwent the most rigorous evaluation since its inception. The Castro case established a now famous three-prong standard for allowing any DNA evidence into a courtroom. The first prong questioned whether there was a generally accepted scientific theory that DNA is unique to the individual. The judge concluded that it is generally accepted in the scientific community that DNA is unique to the individual and that scientists have identified STR and VNTR sequences that vary between individuals with

established allele frequencies in various populations, so prong-1 was accepted in the Castro case. Under the second prong, the court questioned whether the *techniques* used to test DNA could produce reliable DNA results, and concluded that the current PCR-STR and RFLP-VNTR testing techniques are reliable and accurate when performed correctly. The judge stated, “DNA forensic identification tests to determine inclusions are reliable and meet the Frye standard of admissibility” (Patton, 1990). So prong-2 was met in the Castro case. The third prong challenged whether the lab (Lifecodes in this case) performed accepted DNA tests in trial, and found that Lifecodes did not follow accepted scientific procedures and failed to perform certain experiments and use controls. So prong-3 was not met in this case, and the DNA evidence was not allowed at trial. The Court wanted all to be described explicitly with reference to specific characteristics, matching rules, procedures, and statistical probabilities. Due to the fact that DNA testing science had progressed dramatically since the Frye 1923 case, a more methodical approach to reviewing DNA evidence was now necessary.

The ruling was moot, as the Castro case never went to trial, Castro confessed his guilt in late 1989. Following this landmark trial, the Court also determined that a standard protocol of DNA testing needed to be established. So, the FBI created the “Technical Working Group on DNA Analysis Methods” (TWGDAM) to establish universal methods for testing DNA (Patton, 1990). Castro was the first U.S. case where DNA evidence was strongly critically questioned, and provided one of the most detailed dissections of DNA profiling performed at that date in a courtroom.

Two Bulls v. US, 1990

Matthew Sylvester Two Bulls was charged with aggravated sexual abuse of a minor when he raped a fourteen-year-old girl on the Pine Ridge Indian Reservation in South Dakota. Investigators took the victims underwear she was wearing during the attack, and found a semen stain. After comparing Two Bulls' blood DNA profile with the stain profile there was a high probability that he was the attacker (*US Law....1990*). Before his trial, Two Bulls attempted to make the DNA evidence inadmissible in court, but the district judge ruled that DNA evidence was accepted by the scientific community and could be presented to the jury. When approached with a plea agreement, Two Bulls pleaded guilty and was sentenced to nine years in prison followed by two years of supervised release. He did not go to jail right away because he waited on bond for his appeal to go through.

In his appeal, the defense argued that the trial court only applied the Federal Rule of Evidence 702 instead of a more rigorous standard such Frye (*US Law...1990*). The defense argued that the district court violated Two Bulls' due process because the pre-trial evidence hearing was not up to standard, and they should have used a more rigorous Castro standard. The prosecution argued that a Castro hearing is too long and drawn out before trials, whereas using Rule 702 creates a more liberal rule of admissibility which is read differently than the other standards for evidence while still having a proper foundation for scientific testing (*US Law...1990*). The court ruled that Frye and Rule 702 contain different criteria for determining admissibility of forensic evidence which allows them to be combined into a new 5-prong standard. The trial court is to decide whether DNA evidence is *generally accepted* by the scientific community (Frye), whether the testing procedures used in this case are generally accepted as *reliable* if performed correctly (Rule 702), whether the test was performed properly

in this case (Castro), whether the evidence is more prejudicial than probative in this case (Rule 702), and whether the statistics used to determine the probability of someone else having the same genetic characteristics is more probative than prejudicial (Rule 702) and if so disallow it (*US Law*....1990). Thus, the Two Bulls case set a new 5-prong standard that combined three of the previous standards (Frye, Rule 702, and Castro) into one thorough rigorous standard.

Paul Eugene Robinson, 1994, 2000, 2003

The victim in this case, 24-year-old Deborah L., awoke in Sacramento in 1994 to find defendant, a man she had never seen before, standing in her bedroom. He was wearing garden gloves and holding a kitchen knife. When she started to scream, he threatened to kill her if she did not shut up (*People v. Paul Eugene Robinson*, 2000). The unknown male then proceeded to rape her in her second floor apartment. Deborah L. immediately called the police, but he got away. The press dubbed him “The Second Story Rapist” (Delsohn, 2001).

With no leads and the 6-year statute of limitations set to expire on August 24, 2000, the rapist would never be able to be punished if not found before that date. Sacramento Police Detective, Peter Willover, did not want to throw away evidence from the Second Story Rapist because he was still at large and a threat to society. He called Anne Marie Schubert, a sexual assault prosecutor and top DNA expert, for her thoughts on keeping old cases police believed were tied to the same criminal on the map. Ironically she had just been researching a prosecutor in Milwaukee named Norman Gahn (Delsohn, 2001). Gahn had filed a ‘John Doe’ warrant against a suspect in three rapes that would potentially help the police solve several linked unsolved crimes to avoid the statute of limitations. In a John Doe warrant, instead of identifying the suspect by name, birth date, and physical characteristics, the criminal was listed on the warrant by his DNA profile that he had left through the crimes. While the criminal John Doe

remained at large, the statute of limitations had been lifted because a warrant for his arrest had now been served, giving prosecutors more time to put a real name to the John Doe (Delsohn, 2001). Schubert's goal was to apply this same John Doe warrant technique to the Second Story Rapist because police because prosecutors did not want violent serial criminals to get away with crimes because of a statute of limitations.

In November of 1998, four years after committing the Second Story Rapist crime, and two years before the John Doe warrant was filed in 2000, Paul Eugene Robinson a 31-year-old ex con was arrested for violating his parole for an earlier burglary case. Police caught him trespassing and loitering on private property, perhaps looking for his next robbery victim (Delsohn, 2001). Robinson pleaded no contest to the loitering charges, and waited to be sent back to the state prison for violating his parole. Police checking his criminal history believed that Robinson might be responsible for other crimes, and used the then new "DNA and Forensic Identification Data Base and Data Bank Act of 1998" to take his blood and saliva for DNA testing. Robinson's record indicated a previous misdemeanor of spousal abuse, but in California this is a felony, which allowed police to take his DNA and enter his profile into CODIS.

In 2000, three weeks after the John Doe warrant had been served and the unknown Second Story Rapist's profile entered into CODIS, Detective Willover in Sacramento got a call that Robinson's DNA was a cold hit, which is a match between the John Doe assailant's profile and the profile of a person previously entered into CODIS (*People v. Paul Eugene Robinson*, 2000). A new warrant was filed against Robinson, this time under his real name with his real physical description, and he was found hiding at a relative's house.

In Robinson's trial, the use of old DNA using a John Doe warrant would be tested in the court for the first time. Schubert stated that the computer version of her John Doe warrant

directed any law enforcement officer reading it to an additional set of "remarks" that included Robinson's full genetic coding and the admonition to contact Detective Willover. Because of that information, the Judge allowed Robinson to go to court, where his DNA profile was accepted as evidence, and he was sentenced to 65 years in state prison (Delsohn, 2001).

Robinson filed an appeal on the basis that his statute of limitations had expired by the time he was arrested on the John Doe Warrant. On appeal, the defendant argued that the DNA profile on the John Doe warrant did not provide the details of identification of the offender required by section 804, subdivision (d) (*People v. Paul Eugene Robinson*, 2000). Jill Spriggs, an assistant criminal laboratory director for the California Department of Justice, testified that once a John Doe DNA profile is developed, a statistical calculation is performed to determine the frequency of that particular genetic profile in a random unrelated population. The probability of a random match using the 13 locus analysis performed in this case was one in 650 quadrillion in the African American population, one in six sextillion in the Caucasian population, and one in 33 sextillion in the Hispanic population (*People v. Paul Eugene Robinson*, 2000). There are no reported cases of two people matching at all 13 loci, other than identical twins. The defendant's pattern of violent conduct led the court to find that Robinson is a serious danger to society, and the court concluded it had found more than adequate aggravating factors to justify filing the warrant. After the trial court sentenced defendant to 65 years in state prison, the Court of Appeal affirmed the judgment (*People v. Paul Eugene Robinson* 2000). This made Robinson the first person to be convicted of a crime based solely on his DNA evidence.

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CHAPTER-4: DNA DATABASES

Gabrielle Demac

As mentioned in earlier chapters, DNA analysis has grown and developed into one of the most effective crime fighting tools used today (Collins, 2002). With this rapidly growing and improved identification technology, police and investigators have been able to take the smallest bodily fluid or piece of tissue and directly place suspects at the scene of a crime. But when all of this DNA information is gathered, where is it all stored? It's stored in DNA computer databases. But along with these mass DNA information storage centers come many ethical issues. How accurate are the probability estimates of random matches occurring to a database? Whose DNA profiles should be entered into the database? Can someone learn about my medical predispositions from a DNA database? The purpose of this chapter is to introduce the topic of DNA databases, and investigate their ethics.

What is CODIS?

CODIS, also known as the Combined DNA Index System, has become a significant aspect of DNA forensics. Began in 1989, the first database contained only profiles from convicted murderers and sex offenders (U.S Department...2002). But the power and capability of the database to produce fast, accurate results led to more arrests and a stronger acceptance into society. By 2010, CODIS contained over 8,646,417 offender profiles, 328,067 forensic profiles, and aided 119,764 investigations (CODIS Brochure, 2011). Its precious ability to “assist law enforcement by providing investigative information in those cases in which crime scene evidence has yielded a DNA profile while no suspect has been identified” has proven its worth in society (Adams, 2002). This giant computer network is actually divided into 3 levels, or tiers, and

connects the information stored at each level (Figure-1) (U.S Department...2002). The lowest level in CODIS is referred to as LDIS (Local DNA Index System). It consists of local crime labs and police or sheriff's offices that operate at a district level. Any information here is able to be accessed by a DNA system higher in the chain. Above LDIS is the State DNA Index SDIS, which is monitored by assigned agency officials. Each state is assigned a lab to process and store the profiles, while the SDIS maintains communication between the LDIS and National System. The NDIS, national level database, has permission to allow comparison of profiles between state labs since it is maintained by the FBI per DNA Identification Act of 1994 (U.S Department...2002).

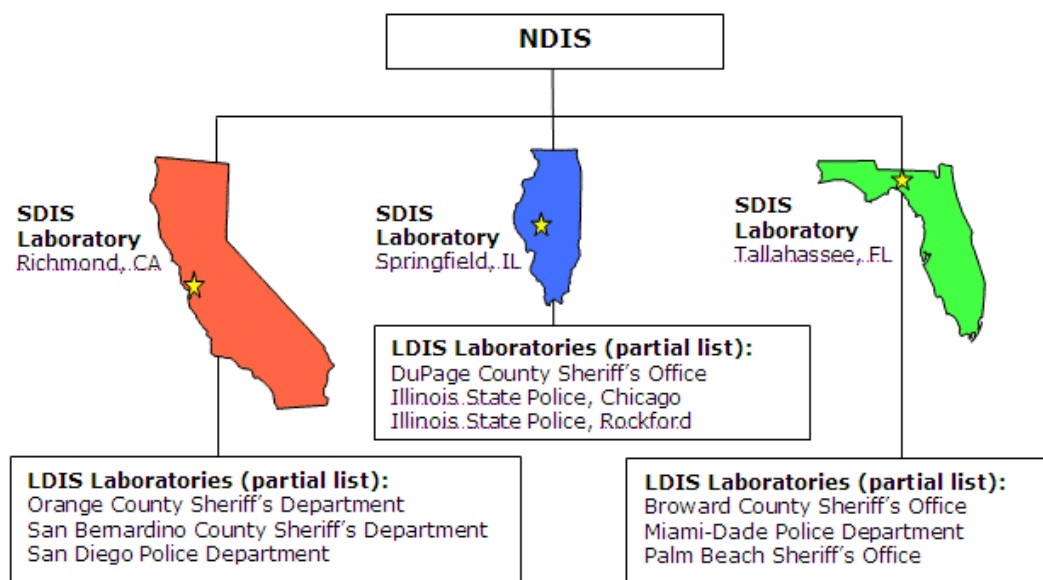


Figure-1: Diagram of the Three Main Tiers of CODIS. CODIS is composed of overlapping local, state, and national tiers which interact to share information on previously convicted individuals and crime scene evidence profiles. (Office, 2006)

So, how does CODIS work? The goal of the database (once the sample backlog is caught up) is to store all DNA profiles found at crime scenes or gathered for other federal purposes, and use them to link crimes and match suspects. Once a DNA profile is developed from crime scene evidence via PCR-STR analysis (as explained in chapter-1), the unknown profile is put in to the forensic index of CODIS and run against the over 8,646,417 offender profiles and 328,067 forensic profiles (2010 numbers) already entered into the database (Adams, 2002; CODIS, 2010). If a match is found in either of the two main indexes of CODIS, the missing person's index or criminal index, there are specific confidentiality procedures that are then followed to confirm the match and hopefully identify the perpetrator (Adams, 2002).

CODIS Versus Genetics Databases

With all of the scientific DNA breakthroughs occurring at such a rapid pace, many may confuse the use and purpose of a forensic database like CODIS with genetics databases used for medical purposes. Although both types of data entries are based on DNA, they are NOT the same. As discussed in Chapter-1, CODIS entries are based on analyzing the 13 core STR loci chosen specifically because they vary widely between individuals and contain no other information. Genetic databases are based on DNA sequencing information, which can include medical predisposition information. CODIS profiling is solely used for identification purposes and there is no medical or personal information within the 13 core STR loci. What is entered into CODIS for a particular locus is the pattern of repeat sequences you have at that location (for example, a pattern of 2 and 4 repeats of the sequence GC at locus-1) (Adams, 2002). Some individuals worry about CODIS being hacked, and the information released to the public. But CODIS contains no medical information.

Genetic databases are not CODIS. These databases are created for the purpose of mapping specific genes and mutations to diseases, or understanding how related two individuals are historically (Bereano, 2000). An example of a genetic database is the Icelandic database currently being used to link genes to diseases (Hlodén, 2000). This database was recently used to identify a mutation in a human gene that prevents people from getting Alzheimer's disease (Jonsson, et al., 2012). The mutation lies within the gene encoding the amyloid precursor protein (APP), and individuals with the mutation produce no amyloid-beta neurotoxin that initiates Alzheimer's disease, even when containing other genes that normally predispose the individual to the disease.

With genetic databases comes a greater risk of loss of privacy, or potential discrimination by medical or life insurance companies who could use the genetic predisposition information to deny an individual medical insurance. Scientists are only now beginning to understand which genes and mutations map to disease predispositions, the analysis so far is crude, but may someday be meaningful. So, individuals contributing their DNA samples to genetic databases should do so only with informed consent. To get the necessary medical predisposition information, someone would actually have to hack into two databases, one containing the DNA sequences, and one linking the DNA entry with personal identifying information. Perhaps the latter information should be kept offline for security purposes, although someone could also steal the list itself.

CODIS Match Probabilities

One key feature CODIS is its ability to help scientists determine more accurately specific allele frequencies at specific loci. When a crime scene DNA profile matches a database

entry, authorities need to know how often a similar match would occur randomly (Brenner, 2004). For example, if an individual is found to have 2 and 4 repeats at locus-1, how frequent is this particular pattern in the general population? Or how frequent is that pattern among Hispanics? To determine this requires an estimate of allele frequencies. The larger the database, the more accurately scientists can calculate the frequency in a given population. Determining that a 2,4 pattern occurs in about 10% of the population is a more accurate statement if several million samples have been screened than 100. If 13 different core loci are analyzed for the total profile, then the frequencies of each of the 13 loci are multiplied together to calculate the overall chance of a random match occurring. When all 13 core loci are analyzed, the chance of a random match occurring is one in several billion. During important criminal cases, such as the OJ Simpson case, scientists sometimes analyze more than 13 loci to further increase the odds of a random match to one in several quadrillion (Brenner, 2004). In order for the data to be accepted in the courtroom, there needs to be strong proof that there is no chance that a given crime scene sample can belong to no one but the suspect; that a random match would be extremely rare. Databases facilitate this process by helping scientists determine how frequent specific patterns are in large populations. The over 8 million profiles currently on record in CODIS helps scientists increase the accuracy of the results (US Department, 2000). During an investigation, if a match occurs to a previous offender, investigators then obtain a warrant for a new DNA sample from the suspect to perform a more rigorous test for confirmation and to obtain stronger evidence to be used in court (US Department, 2000).

Whose DNA Gets Entered?

When CODIS was first created, only the profiles of convicted murderers and sex offenders were required to be entered. Then as time progressed, the federal government required any offender “convicted of federal or military crimes” to add their profiles, and soon after that, any felon or missing person were added (US Department, 2000). Federal law requires all states to enter profiles into CODIS, but each individual state determines whose profiles are entered. Some states require only individuals convicted of violent or sex crimes to be entered, while other states allow profiles from *arrested* individuals, despite the constitutional presumption of innocence (Bereano, 2000).

From purely a crime solving perspective, *everyone*’s DNA should be included in CODIS not just previous offenders, as this would increase our chances of solving new crimes. This could be achieved in the future by taking a cheek swab at time of birth. However, due to privacy rights (discussed below) no state currently requires this. The state of Massachusetts currently requires *convicted* felons and some *convicted* juveniles to submit their DNA profiles. Only 15 states currently require *arrestees* to submit their DNA samples (National Conference of State Legislatures, 2010).

Potential Limitations

CODIS became so successful at helping authorities solve crimes there is a huge backlog of DNA samples awaiting analysis. This backlog is a serious problem for crimes nearing their 6-year statute of limitations. In 2000, Congress passed the ‘DNA Analysis Backlog Elimination Act’ (DNA Analysis...2000) to attempt to help alleviate the sample backlog problem. However,

this was followed in 2001 by the USA Patriot Act which expanded the list of offenses allowed in CODIS, which further increased the backlog (USA, 2001; Office, 2006). Unanalyzed samples in the forensic index, the convicted offender index, and other evidence such as unprocessed rape kits, all contribute to the growing backlog, requiring labs to prioritize cases (U.S. Department, 2000). Although with the FBI's support program, CODIS has flourished, and many states have further increased the types of crimes requiring entries.

Database Ethics and Privacy Rights

One of the largest concerns with CODIS is personal privacy. Is the data being entered really accurate? Who has access to this information, and when? Are the people and computers storing all of this information faithful/safe/carefully monitored? Many questions arise when any form of a person's identity is involved.

As mentioned before, unlike many other branches of genetic research and medical databases, no genetic predisposition data is entered into CODIS. The information entered includes the DNA profile of 13 core loci, the name of the laboratory making the analysis, the specimen ID number, and the agency submitting the profile (Adams, 2002). However, even if CODIS does not contain sensitive genetic information, its entries are still held confidential. According to the DNA Identification Act of 1994, all DNA information will be held confidential, and any disclosure of information given without permission will result in a penalty of up to \$250,000 (Adams, 2002). Any computer containing CODIS is securely stored at an assigned criminal justice agency, and the only people with access to this data are CODIS-authorized and FBI-approved agents, and occasionally defendants (if the data has a direct connection to their case) (Adams, 2002).

In an attempt to ensure the public's privacy rights are being upheld, many states constantly make changes, repeals, and amendments to existing laws to adjust to new advancements in DNA technology. Some states have enacted laws extending or eliminating the statute of limitation for some crimes. This increases the length of time that some entries are held, increasing the chance of a breach of information (U.S. Department, 2000). The longer the information is stored, the more potential there is for someone to obtain it. As discussed in Chapter-3 on landmark DNA cases, the Paul Eugene Robinson case in 2000 established precedence for allowing John Doe warrants. These special warrants for an individual's arrest contain only a DNA profile from crime scene evidence (which is the only thing known about the person's identity at that time), without stating the individual's name or physical description as traditionally appear on a warrant. Filing a John Doe warrant for that "person" arrest effectively stops the clock on the 6-year statute of limitations. The DNA profile itself is listed as the criminal. This allows cold cases to remain active for longer periods, and the "john doe" DNA to be used in court once matched to an actual person (U.S. Department, 2000).

Chapter-5 Conclusions

One of the greatest concerns about DNA databases are privacy rights. According to an article written in 2000 by Phillip Bereano, "Although new technologies claim to offer us more 'freedom,' they really can threaten our civic values. Beyond the risks of discrimination and loss of privacy, however, society's fascination with genetics determinism has other social and political consequences." In regards to DNA databases such as CODIS and privacy rights, what many people fail to realize is there is no personal information or medical information entered in CODIS. There are far more benefits to using the database than there are legitimate risks. The

most important arguments on CODIS instead relate to who should be required to provide their DNA sample. While the “FBI has been promoting genetic screening of criminals to establish state DNA identification data banks to be used in criminal investigation” (Bereano, 2000), some states have been establishing laws requiring even just those *accused* of a crime to submit a profile. CODIS was not designed to discriminate or publicly display personal information, but to compare target DNA records found as evidence to previous entries in the database, to help determine allele frequencies more accurately, and to help authorities solve crimes (Adams, 2002). When a match is found, only the necessary people involved in the investigation are informed, not even the FBI or local lab are notified unless it directly pertains to them (Adams, 2002). DNA databases allow for cross-comparison of DNA profiles between local, state, and national levels to help link crimes and possibly catch criminals faster. By allowing for cross-comparisons, if a match is found different states or jurisdictions are then capable to coordinate investigations and solve crimes more efficiently (U.S. Department, 2000). In an attempt to ensure the privacy and accuracy of the information being stored in the databases, recommendations, procedures, and routine audits (Table-I) have been developed to help manage and monitor the accuracy and efficient use of CODIS, analyze the information discovered through that database’s use, and examine the actions following these discoveries (Office, 2006).

Table I: Safeguards and Audits for CODIS.

Recommendations to FBI:

- Require that the accuracy, completeness, and allowability of the DNA profiles in the national index be routinely verified through audits or other means.
- Ensure that analysts performing DNA testing at laboratories uploading DNA profiles to the national index are aware of the NDIS requirements, particularly those requirements delineating the types of allowable profiles.
- Develop and implement a process to ensure that laboratories adequately resolve all deficiencies noted during the QAS-required audits.

This audit was designed to determine the present status of CODIS operations.

To accomplish these objectives, we reviewed various data and documentation provided to us by FBI officials, evaluated the results of past OIG CODIS laboratory audits, interviewed members of the CODIS Unit staff, and collected documentation from select NDIS-participating laboratories to analyze:

- CODIS unit staffing and responsibilities;
- the accuracy of NDIS Audit Review Panel (Review Panel) records;
- the timeliness of the Review Panel process;
- CODIS program goals, objectives, and measurements;
- CODIS unit oversight and monitoring of participants;
- weaknesses in compliance with QAS or NDIS participation requirements;
- the adequacy of the FBI's corrective actions to our previous recommendations;
- the FBI's implementation of legislated changes to NDIS; and
- the FBI's management of CODIS operations and infrastructure.

(Office, 2006)

Chapter-5 Bibliography

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PROJECT CONCLUSIONS

Since its initial development in 1985, DNA fingerprinting is a tool that has grown and proven its worth to forensic scientists and the criminal justice system over the years. DNA, although 99.8% identical between all humans, has been used by scientists to identify individuals based on the analysis of 13 core loci using techniques such as RFLP and STR-PCR. RFLP type fingerprints were the first developed. This procedure requires a relatively large amount of DNA and can take a few weeks to perform, but is less prone to contamination effects than other techniques. More recently, the STR-PCR technique is used most frequently due to its speed and sensitivity. The use of PCR allows scientists to rapidly amplify very small amounts of DNA from trace forensic samples. Even though there is a higher risk of contamination, STR-PCR techniques are more commonly used today because of the small samples size used and rapid results produced.

Using the best STR-PCR or RFLP type analyses is totally useless unless the DNA is properly collected, transported, and stored prior to analysis. DNA forensics has become such a significant tool to scientists and law enforcement authorities, that with its growth has come advancements in the proper collection and storage methods used to preserve and prevent any degradation to these samples. Carefully controlling the crime scene to prevent evidence contamination or degradation, and maintaining a Chain of Custody to ensure only authorized individuals handle the evidence, helps DNA evidence gain acceptance in courts.

In order to enter complex DNA evidence in U.S. courts, several landmark court cases have set legal precedence to require pre-trial hearings to ensure the tests were properly run, and to help standardize the technology to further facilitate its acceptance in courts. Currently,

through the *Two Bulls vs. US* court case, the courts have decided upon a five-prong standard that combines three previously accepted standards: Frye, Rule 702, and Castro. During a pre-trial court hearing, the judge uses a five-prong standard to determine whether the DNA technology used is *generally accepted* by the scientific community (Frye), whether the testing procedures are generally accepted and *accurate* when performed correctly (Rule 702), whether the testing has been completed using standardized procedures in the case (Castro), whether the evidence is more prejudicial than probative (Rule 702), and whether the statistics used to find the probability is more probative than prejudicial (Rule 702). Combining all three standards has given the courts a thorough rigorous basis for allowing DNA evidence into the courtroom.

After investigating the use and ethics behind DNA databases, we feel as though most US citizens should have their DNA entered into the CODIS system. Realizing that doing this would be highly controversial, by having more people's DNA profiles entered into CODIS, it would allow matches to be made to first time offenders instead of only to repeat offenders, and its larger size would further increase the accuracy of assigning allele frequencies to the 13 core loci. Although hacking into a medical genetics database could provide information on medical predispositions (if the same database contained individual identifying information), these privacy concerns are far fewer for CODIS that enters only STR repeat information for core loci of junk DNA. CODIS procedures strictly mandate the entry of only non-identifying information, and access is limited only to authorized individuals; thus there is little reason to fear a privacy breach or medical predisposition information being obtained from this type of database. However, because everyone's DNA profile would be in the database, including totally innocent people, authorities would have to use extreme caution when a match is potentially found to the database to ensure innocent people are treated fairly until corroborating proof is obtained.